CHES STUDIES

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NORTH CAROLINA BIRTH DEFECTS UPDATE

by

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ABSTRACT

The North Carolina Birth Defects Registry is an important source of data for the ongoing surveillance of birth defect incidence in the state. This study presents an update on the Birth Defects Registry using 1988 data. This was the first year of using an expanded birth certificate which includes a new method of recording birth defects as well as some new items relating to medical characteristics of the mother and child. Selected tabulations of the 1988 data are presented and incidence data for forty-four of the most common birth defect conditions are reported. Future Registry modifications are also discussed. Use of Birth Defects Registry data by health and environmental science researchers and public health planners is encouraged and supported by the State Center for Health and Environmental Statistics (SCHES). Special tabulations of the data are available by request.

INTRODUCTION

The North Carolina Birth Defects Registry has documented congenital anomalies and other birth disorders in North Carolina since 1984. A comprehensive report on the development of the Birth Defects Registry and the 1984-86 Registry data analysis is the CHES Studies report, "North Carolina Surveillance of Birth Defects" (1). The present report provides an update on the incidence of birth defects in North Carolina using 1988 data.

According to the Registry data, birth defects are found in about five percent of North Carolina live births. While some birth defects are obvious at birth, many are not readily detected at or near the time of birth. For this and other reasons, complete ascertainment of birth defects is not possible using birth certificates only. The Birth Defects Registry is a combination of separate and distinct sources of data, thereby providing a more complete account of birth defects in North Carolina.

The terms "birth defect" and "congenital anomaly" are often used interchangeably. But to be more specific, congenital anomalies are conditions at birth that have diagnosis codes of 740.0-759.9 in the International Classification of Diseases, ninth revision. Several other newborn conditions are captured in the Registry which may be classified as birth defects, e.g., various endocrine and metabolic disorders, some anemias, cerebral palsy and other selected central nervous system disorders, fetal alcohol syndrome, and certain infections.

The Registry provides for relatively acccurate reporting of congenital anomalies because it is created by merging several different data files with birth and infant death records. Identification of birth defects after the newborn period (the first week of life) is improved. More accurate birth defect statistics can enhance the efforts of public health planners, epidemiologists, toxicologists, and others who study the causes and implications of birth defects.

In 1988, an effort was made to improve reporting of congenital anomalies on birth certificates. The birth certificate form now has check boxes in the congenital anomaly section instead of "fill-in-the-blank" (open-ended) reporting fields (see **Appendix A**). Congenital anomalies listed on the revised birth certificate include the anomalies most prevalent in North Carolina. The implementation of the new birth certificate format has resulted in somewhat better reporting. The percentage of all births which have a congenital anomaly reported on the birth certificate has increased from 0.8 for the 1984-86 period to 1.1 for 1988. The birth certificate data alone, however, still provide a very incomplete description of birth defects in North Carolina.

METHODS

A number of agencies that maintain computerized databases which include specific data on birth defects have been essential contributors to the North Carolina Birth Defects Registry. The birth and infant death records maintained by SCHES serve as the starting point and base for the Registry.

Linked to these records are newborn Medicaid claims records, newborn hospital discharge records, and Children's Special Health Services (CSHS) records obtained by SCHES. Once the linkage is completed, each Registry data record consists of birth certificate information and birth defect diagnoses (if found) from one or more of the data sources previously described.

The CSHS program serves poverty-level children with various developmental disabilities who are treated through the public health department system. With this data set, provided by the Health Services Information System (HSIS), birth defects detected and treated after the newborn period can be identified. Of the 901 total CSHS records for persons born in 1988 that had a birth defect indicated, 797 (88%) were matched to 1988 birth certificates.

Professional Activity Study (PAS) and other abstracting systems were used to acquire 1988 hospital discharge data for newborns. Much time and effort were needed to incorporate the hospital discharge data into the Birth Defects Registry. Cumbersome hand-matching of hospital discharge records to birth records, in addition to computerized matching, was necessary to make full use of these data files. A more complete description of the methods used to create the Birth Defects Registry can be found in the previous report on birth defects (1).

RESULTS

The Registry indicates that there were 5,272 births in 1988 with at least one type of birth defect diagnosis (5.4% of all births). Of all births, 4,420 or 4.5% were reported to have at least one congenital

anomaly (ICD-9 codes 740.9-759.9 only). As shown in **Table 1**, the newborn hospital discharge records identified the most congenital anomalies for the Registry. Newborn Medicaid claims records provided the second largest number of congenital anomaly diagnoses; birth certificate records provided the third largest number of congenital anomalies.

TABLE 1
Numbers of Congenital Anomalies Reported by Source of Data and Percent of all Births North Carolina, 1988

Source of Data	Number of Records	Percent of All Births*
Newborn Hospital Discharge Records	2,488	2.6
Newborn Medicaid Claims	1,189	1.2
Birth Certificates	1,043	1.1
Children's Special Health Services Records	740	0.8
Infant Death Certificates	256	0.3

*Total number of resident live births in 1988 = 97,560

One can see the problem of solely using birth certificates, or any one of the other data sets alone, to determine birth defect incidence in the state. Only 1.1 percent of 1988 births were reported to have congenital anomalies according to birth certificates. This is only one-fourth of the total incidence shown by the Registry **(see Table 2)**. Likewise, each of the other data sets alone does not yield birth defect statistics which represent the actual size of the problem.

TABLE 2
Comparison of Birth Certificate Reported Congenital Anomalies with Birth Defect Registry Reported Congenital Anomalies North Carolina, 1988

ICD-9 Code	Birth Certificate Number	Birth Certificate Percent	Registry Number	Registry Percent
740-759	1,043	1.07	4,420	4.53

An analysis of the unique contribution of each of the data sets to the Registry **(see Table 3)** shows that 34.6 percent of the congenital anomalies are provided exclusively by hospital discharge records. It is expected that this percentage would have been even higher had the hospital discharge data for newborns been geographically complete. In 1988, about 30 percent of the state's hospitals did not have accessible data for Birth Defects Registry purposes. It is for this reason that a geographical study of birth defects, at this point, would not be valid. Geographical areas showing a low incidence of malformations, for instance, would more than likely be areas for which some of the hospital discharge data are missing.

TABLE 3
Numbers and Percentages of Congenital Anomalies Reported by Unique and Multiple Data Sources North Carolina, 1988

Source of Data	Number of Records	Percentage
Sole Source		
Newborn Hospital Discharge Records	1,530	34.6
Children's Special Health Services Records	675	15.3
Birth Certificates	517	11.7
Newborn Medicaid Claims	490	11.1
Infant Death Certificates	92	2.1
Multiple Sources of Data	1,116	25.2
Total	4,420	100.0

It is also possible that some birth defect diagnoses may be missing on the computerized hospital discharge records in North Carolina as compared to diagnoses recorded in the hospital medical records. A study by Calle and Khoury (2) revealed that a significant number of birth defects were missing on computerized hospital discharge records, especially for those births with multiple defects. Review of medical records indicated that 6.9 percent of the births had at least one congenital anomaly; the hospital discharge records showed only 3.5 percent.

The 1988 newborn Medicaid claims files identified many more congenital anomalies than the 1984-86 claims files. For the 1984-86 period, newborn Medicaid claims records uniquely supplied only 1.6 percent of the Registry's cases. In 1988, those records uniquely supplied 11.1 percent of the Registry's cases **(see Table 3)**. Part of the reason for this large difference is

a change in the Medicaid eligibility policy. In 1988, many more pregnant women and their babies became eligible for Medicaid benefits due to legislated expansions of the program.

Table 4 gives numbers of cases of specific defects and birth disorders listed under several broad categories (see **Appendix B** for definitions). These are numbers of the selected birth defects reported in the entire 1988 Registry. In descending order, the following types of defects were the most frequently reported in the state: other musculoskeletal/integumental anomalies (1,754), malformed genitalia (562), clubfoot (434), heart malformations (403), other

TABLE 4

Numbers of Cases of Selected Birth Defects (With ICD-9 Codes)
North Carolina, 1988

	Number
CENTRAL NERVOUS SYSTEM	
Anencephalus (740)	16
Spina Bifida/Meningocele (741, 742.0)	57
Hydrocephalus (742.3)	69
Microcephalus (742.1)	25
Other Central Nervous System Anomalies (742.2, 742.4-.9)	33
EYE	
Anophthalmos/Microphthalmos (743.0, 743.1)	8
Congenital Cataract (743.3)	4
CARDIOVASCULAR	
Heart Malformations (Total) (745, 746)	403
Common Truncus (745.0)	0
Transposition of Great Arteries (745.1)	12
Tetralogy of Fallot (745.2)	7
Ventricular Septal Defect (745.3, .4, .7)	93
Atrial Septal Defect (745.5)	24
Endocardial Cushion Defect (745.6)	7
Anomalies of Pulmonary Valve (746.0)	16
Tricuspid Valve Atresia and Stenosis (746.1)	1
Aortic Valve Stenosis and Atresia (746.3, .4)	5
Hypoplastic Left Heart Syndrome (746.7)	22
Other Circulatory/Respiratory Anomalies (747, 748)	388
Patent Ductus Arteriosus (747.0)	193
Coarctation of Aorta (747.1)	3
Pulmonary Artery Anomaly (747.3)	12
Agenesis, Hypoplasia, or Dysplasia of the Lung (748.5)	59
OROFACIAL	
Cleft Lip/Palate (749)	120
Cleft Palate without Cleft Lip (749.0)	35
Cleft Lip with and without Cleft Palate (749.1, .2)	42
GASTROINTESTINAL	
Rectal Atresia/Stenosis (751.2)	31
Tracheo-esophageal Fistula/Esophageal Atresia (750.3)	16

	Number
GASTROINTESTINAL *(con't)*	
Omphalocele/Gastroschisis (756.7)	57
Other Gastrointestinal Anomalies (750.0-.2, 750.4-.9, 751.0, .1, 751.3-.9	91
GENITOURINARY	
Malformed Genitalia (752)	562
Renal Agenesis (753.0)	23
Other Urogenital Anomalies (753.1-.9)	79
MUSCULOSKELETAL	
Polydactyly/Syndactyly/Adactyly (755.0, .1)	373
Clubfoot (754.5-.7)	434
Diaphragmatic Hernia (756.6)	41
Other Musculoskeletal/Integumental Anomalies (754.0-.4, 754.8, 755.2-.9, 756.0-.5, 756.8-.9, 757.0-.9)	1,754
CHROMOSOMAL	
Down's Syndrome (758.0)	65
Other Chromosomal Anomalies (758.1-.9)	44
Trisomy 13 (758.1)	8
Trisomy 18 (758.2)	16
INFECTIONS	
Congenital Syphilis (090)	11
Gonococcal Infections (098)	3
Congenital Rubella (771.0)	0
METABOLIC	
Cystic Fibrosis (277.0)	4
Phenylketonuria (270.1)	5
OTHER CENTRAL NERVOUS SYSTEM DISORDERS	
Infantile Cerebral Palsy (343)	39
Epilepsy (345)	11
OTHER CONDITIONS	
Congenital Hypothyroidism (243)	5
Coagulation Defects (286)	14
Fetal Alcohol Syndrome (760.71)	47
Hereditary Hemolytic Anemias (282)	22
TOTAL INFANTS HAVING BIRTH DEFECTS	**5,272**

circulatory/respiratory anomalies (388), and polydactyly/syndactyly/adactyly (373). Birth defect rates (i.e., percent of all births with these defects) were not computed since the data in Table 4 are for only one year. Rates or percentages based on fewer than 20 cases in the numerator may have a large random error and should not be used for comparison purposes.

Table 5 contains a subset of the conditions listed in Table 4. Frequencies of the congenital anomalies captured by the new birth certificate check boxes are compared to those captured in the total Registry. The numbers in the Total Registry column are the same as those found in Table 4. The Birth Certificate column contains the numbers of cases identified by reviewing birth certificate records alone. The ratios indicate the proficiency of birth certificate reporting of congenital anomalies compared to the Registry as a whole. Birth certificates captured only eight percent of certain circulatory and respiratory anomalies, for example. On the other hand, some of the other conditions (e.g., anencephalus and cleft lip/palate) are relatively well reported on birth certificates.

TABLE 5

Comparison of Birth Certificate Congenital Anomaly Check Box Items to Total Registry Results
North Carolina, 1988

Congenital Anomaly	Birth Certificate	Total Registry	Ratio
Anencephalus	12	16	.75
Spina Bifida/Meningocele	34	57	.60
Hydrocephalus	41	69	.60
Microcephalus	8	25	.32
Other Central Nervous System Anomalies	11	33	.33
Heart Malformations	108	403	.27
Other Circulatory/Respiratory Anomalies	32	388	.08
Rectal Atresia/Stenosis	16	31	.52
Tracheo-esophageal Fistula/Esophageal Atresia	7	16	.44
Omphalocele/Gastroschisis	25	57	.44
Other Gastrointestinal Anomalies	22	91	.24
Malformed Genitalia	137	562	.24
Renal Agenesis	9	23	.39
Other Urogenital Anomalies	30	79	.38
Cleft Lip/Palate	92	120	.77
Polydactyly/Syndactyly/Adactyly	150	373	.40
Club Foot	80	434	.18
Diaphragmatic Hernia	16	41	.39
Other Musculoskeletal/Integumental Anomalies	225	1,754	.13
Down's Syndrome	38	65	.58
Other Chromosomal Anomalies	15	44	.34

In addition to the change in the congenital anomaly reporting system on birth certificates, some new items have been added to the birth certificate. Medical risk factors, tobacco use, and method of delivery (e.g., C-section) are now recorded. **Table 6** shows the above-mentioned birth certificate variables along with age of mother, gender of child, birthweight, and other characteristics as they relate to reported congenital anomalies. Of the births involving at least one pregnancy-related medical risk, 5.6 percent were born with one or more identified congenital anomalies. Fewer congenital anomalies (4.2 percent) were identified among births with no medical risk(s) during pregnancy. Mothers who smoked or who had a C-section were somewhat more likely to have a child with a congenital anomaly. Among Medicaid births, 8.4 percent had one or more recorded congenital anomalies, compared to 3.3 percent for non-Medicaid births. The incidence for non-Medicaid births may be understated here since there was no supplemental data for non-Medicaid births that is comparable to the data on claims paid by Medicaid.

As was found in the study of 1984-86 birth defects, factors associated with a higher incidence of congenital anomalies are young age of mother, male child, nonwhite race, low education of mother, low birthweight, and especially infant death. The differences in the percentages for each of the items in Table 6 were found to be statistically significant ($p<.001$), except for smoking.

TABLE 6

Percentages of Births with One or More Congenital Anomalies by Selected Mother and Child Characteristics North Carolina, 1988

Characteristic		Percentage of Births with Anomalies
Age of Mother	Less than 18 years	6.8
	18-34 years	4.4
	35 years and over	4.3
Gender of Child	Male	5.0
	Female	4.1
Race	White	3.3
	Nonwhite	7.1
Education of Mother	Less than 9 years	6.0
	9-11 years	6.1
	12 years	4.6
	More than 12 years	3.4
Birthweight	Less than 1500 grams	13.0
	1500-2499 grams	7.7
	2500 grams and over	4.2
Infant Death	Death within 1 year	27.0
	Survived first year	4.3
Medical Risk Factors	Yes	5.6
	No	4.2
Smoked During Pregnancy	Yes	4.8
	No	4.6
C-section	Yes	5.4
	No	4.3
Medicaid Patient	Yes	8.4
	No	3.3

DISCUSSION AND CONCLUSIONS

The North Carolina Birth Defects Registry is compiled annually to provide accurate, detailed data on the occurrence of birth defects throughout the state. These data should be useful to researchers and to those involved in health service planning.

Even though the Birth Defects Registry can be used to provide valuable descriptive and detailed statistics on birth defects in the state, it is not without limitations. Many birth defects still escape identification. Fetal deaths with birth defects, for example, are not accounted for in this Registry. Another problem involves the difficulty in tracking abnormal conditions that become obvious later in a child's life. The Registry captures primarily abnormal conditions that are detectable in the newborn period. The only data sources that identify anomalies that become apparent later are the infant death and CSHS records. Some of the CSHS records are for children up to two or three years of age.

The higher incidence of congenital anomalies among nonwhites and young mothers may be exaggerated due to the contribution of CSHS and Medicaid data to the Registry. A 1988 study, however, by the Centers for Disease Control (CDC) (3), found that even though nonwhite children experienced fewer major and multiple malformations than whites, their overall rate of birth defects was higher. Males have a higher incidence of congenital anomalies than females because male births are associated with malformed genitalia more than female births (1). Infants of older mothers may also have a lower rate of birth defects than infants of younger mothers due to the more extensive diagnostic testing procedures used for older pregnant women which may result in termination of pregnancy.

There has been a considerable delay between the collection of data and completion of the Registry on a yearly basis. This has been primarily due to the time and difficulty involved in acquiring and preparing the hospital discharge data. For the 1989 and subsequent registries, the hospital discharge data will be obtained from the Medical Database Commission. The Medical Database Commission is a centralized hospital discharge data source with the ability to provide more complete, and presumably more timely, data than PAS and the other abstracting services could. The 1989 Registry will also incorporate selected records from the Neonatal Intensive Care Unit (NICU) statewide database. The data are maintained by the University of North Carolina at Chapel Hill and have potential to improve the diagnostic specificity and completeness of the Registry. Another possible source of data in the future is the Medical Genetics Centers records.

The North Carolina Birth Defects Registry is the most complete source of data on congenital anomalies and other abnormal birth conditions that currently exists in the state. Researchers can use the detailed data to help determine associations between specific anomalies and factors such as the physical or occupational environment. Special requests for Registry data are welcomed at SCHES and are not limited to the type of statistics tabulated in this publication. The 1989 Registry data will be available in the near future for data analysis and requests. Progress continues to be made due to the ongoing cooperative efforts of SCHES, the Medical Database Commission, Neonatal Intensive Care Units, the North Carolina Medical Genetics Association, and the Children's Special Health Services program.

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(3) Chavez, Gilberto F., Cordero, Jose F., and Becerra, Jose E., "Leading Major Congenital Malformations Among Minority Groups in the United States, 1981-1986," Morbidity and Mortality Weekly Report Surveillance Summaries, July 1988.

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APPENDIX A

Birth Certificate Congenital Anomalies in Check Box Format (North Carolina, 1988)

CONGENITAL ANOMALIES OF CHILD
(Check all that apply)

Anencephalus ..... 01 ☐
Spina bifida/Meningocele ..... 02 ☐
Hydrocephalus ..... 03 ☐
Microcephalus ..... 04 ☐
Other central nervous system anomalies

(Specify) _____ 05 ☐

Heart malformations ..... 06 ☐
Other circulatory/respiratory anomalies

(Specify) _____ 07 ☐

Rectal atresia/stenosis ..... 08 ☐
Tracheo-esophageal fistula/Esophageal atresia ..... 09 ☐
Omphalocele/Gastroschisis ..... 10 ☐
Other gastrointestinal anomalies

(Specify) _____ 11 ☐

Malformed genitalia ..... 12 ☐
Renal agenesis ..... 13 ☐
Other urogenital anomalies

(Specify) _____ 14 ☐

Cleft lip/palate ..... 15 ☐
Polydactyly/Syndactyly/Adactyly ..... 16 ☐
Club foot ..... 17 ☐
Diaphragmatic hernia ..... 18 ☐
Other musculoskeletal/integumental anomalies

(Specify) _____ 19 ☐

Down's syndrome ..... 20 ☐
Other chromosomal anomalies

(Specify) _____ 21 ☐

None ..... 00 ☐
Other *(Specify)* _____ 22 ☐

APPENDIX B

BIRTH DEFECT DEFINITIONS

[Adapted from Reference (4)]

Adactyly — absence of digits on the hand or foot.

Anencephaly — absence of the cranial vault, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull.

Anophthalmia — complete absence of the eyes (rare) or the presence of vestigial eyes.

Aortic Atresia — absence or closure of the aortic root orifice, where the left ventricle is hypoplastic or nonfunctioning, oxygenated blood passing from the left into the right atrium through a septal defect, and the mixed venous and arterial blood passing from the pulmonary artery to the aorta by way of a patent ductus.

Aortic Stenosis — a narrowing of the aortic orifice of the heart or of the aorta itself.

Artrial Septal Defect — persistent patency of the atrial septum due to failure of fusion between either the septum secundum or the septum primum and the endocardial cushions.

Cerebral Palsy — a persisting qualitative motor disorder appearing before the age of three years, due to a nonprogressive damage to the brain.

Cleft (Facial, oblique) — a rare form of facial cleft extending from the lip to the inner canthus of the eye. It may be superficial but usually separates the underlying bone and is associated with cleft lip, cleft palate, or lateral facial cleft.

Clubfoot (talipes) — a foot that is twisted out of shape or position.

Coarctation of Aorta — segmental or localized narrowing of the lumen of the aorta.

Common Truncus (truncus arteriosus, persistent) — characterized by a single arterial trunk arising from the heart, receiving blood from both ventricles and supplying blood to the coronary, pulmonary, and systemic circulations; sometimes classified by the arrangement of the arteries supplying the lungs.

Congenital Cataract — opacities present at birth; may or may not impair vision. 35-50% of the cases are sporadic and of unknown cause; 25% are inherited; 25-40% are due to intrauterine infection, drug-induced toxicity, ionizing radiation, trauma, prematurity, and chromosomal, endocrine, metabolic, and systemic disorders. Congenital cataracts are often associated with low birth weight, central nervous system abnormalities, mental retardation, convulsions, and cerebral palsy.

Congenital Rubella Syndrome — Transplacental infection of the fetus as a result of maternal infection in the first trimester; can cause death of the conceptus or severe developmental abnormalities in the newborn infant.

Cystic Fibrosis — an autosomal recessive disorder of infants, children, and young adults in which there is widespread dysfunction of the exocrine glands; characterized by signs of chronic pulmonary disease (due to excess mucus production in the respiratory tract), pancreatic deficiency, abnormally high levels of electrolytes in the sweat, and occasionally by biliary cirrhosis.

Diaphragmatic Hernia — a defective development of part of the diaphragm that frequently allows protrusion of abdominal or retroperitoneal structures into the thorax.

Down's Syndrome — a chromosomal disorder characterized by a small, anteroposteriorly flattened skull, short, flat-bridge nose, epicanthal fold, short phalanges, widened spaces between the first and second digits of hands and feet, and moderate mental retardation, with Alzheimer-like disease developing in the fourth or fifth decade. The chromosomal aberration is trisomy of chromosome 21 associated with later maternal age. Called also trisomy 21 and nondisjunction; formerly called mongolism.

Endocardial Cushion Defect — any defect in development of the endocardial cushions (elevation on the atrioventricular canal of the embryonic heart, which later fuses with the free edge of the septum primum to separate the right and left atrial); may include ostium primum ASD with mitral regurgitation or a complete A-V canal; associated with anomalous conduction system; resulting in characteristic EKG findings.

Epilepsy — paroxysmal transient disturbances of brain function that may be manifested as episodic impairment or loss of consciousness, abnormal motor phenomena, psychic or sensory disturbances, or perturbation of the autonomic nervous system. Symptoms are due to paroxysmal disturbance of the electrical activity of the brain.

Fetal Alcohol Syndrome — a pattern of altered prenatal growth and morphogenesis occurring in infants exposed to high levels of alcohol during pregnancy; it includes maxillary hypoplasia, prominence of the forehead and mandible, short palpebral fissures, microphthalmia, epicanthal folds, growth retardation, mental retardation, and microcephaly.

Gastroschisis — a congenital disruption of the abdominal wall not involving the site of insertion of the umbilical cord, and usually accompanied by protrusion of abdominal contents.

Gonococcal Infections — infections caused by the organism causing gonorrhea; transmitted by contact with infected exudates in neonatal children at birth, or by infants in households with infected inhabitants.

Hereditary Hemolytic Anemias (Nonspherocytic) — a heterogeneous group of congenital hemolytic anemias characterized by absence of spherocytosis, negative antiglobulin tests, and absence of a detectable abnormal hemoglobin.

Hydrocephaly — dilation of the cerebral ventricles, most often occurring secondarily to obstruction of the cerebrospinal fluid pathways and accompanied by an accumulation of cerebrospinal fluid within the skull; typically characterized by enlargement of the head, prominence of the forehead, brain atrophy, mental deterioration, and convulsions; may be congenital or acquired and may be of sudden onset or be slowly progressive.

Hypoplastic Left Heart Syndrome — incomplete development or underdevelopment of the left heart.

Hypothyroidism — deficiency of thyroid gland activity; characterized by a decrease in basal metabolic rate, lethargy, linear growth retardation, constipation, and neonatal hyperbilirubinemia.

Microcephaly — abnormally small head circumference, usually associated with mental retardation.

Omphalocele — protrusion, at birth, of part of the intestine through a large defect in the abdominal wall at the umbilicus, the protruding bowel being covered only by a thin transparent membrane composed of amnion and peritoneum.

Patent Ductus Arteriosus — abnormal persistence of an open lumen in the ductus arteriosus after birth, the direction of flow being from the aorta to the pulmonary artery, resulting in recirculation of arterial blood through the lungs.

Phenylketonuria (PKU) — hyperphenylalaninemia, type I: phenylalanine accumulation resulting in mental retardation (phenylpyruvic oligophrenia), neurologic manifestations (including hyperkinesia, epilepsy, and microcephaly), light pigmentation, eczema, and a mousy odor, unless treated by a diet low in phenylalanine.

Polydactyly — the presence of supernumerary digits (extra fingers and/or toes).

Renal Agenesis — failure of development of the kidneys.

Spina Bifida — defective closure of the bony encasement of the spinal cord, through which the cord and meninges may protrude.

Syndactyly — persistence of the webbing between adjacent digits, so they are more or less completely attached; frequently an isolated autosomal dominant trait.

Tetralogy of Fallot — a combination of congenital cardiac defects consisting of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta so that it overrides the interventricular septum and receives venous as well as arterial blood, and right ventricular hypertrophy.

Tracheo-esophageal Fistula — an abnormal passage or communication between the trachea and the esophagus.

Transposition of Great Arteries (of Great Vessels) — the aorta arises entirely from the right ventricle and the pulmonary artery from the left ventricle — life then depends on a crossflow of blood between blood in the right heart and that in the left heart, as through a ventricular septal defect or a patent duct arteriosus. Cyanosis is the chief symptom.

Tricuspid Valve Stenosis — narrowing or stricture of the tricuspid valve.

Trisomy — the presence of an extra chromosome of one type in an otherwise diploid cell ($2n + 1$).

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